Your Guide to Understanding Genetic Conditions

SAA1 gene

serum amyloid A1

Normal Function

The *SAA1* gene provides instructions for making a protein called serum amyloid A1. This protein is made primarily in the liver and circulates in low levels in the blood. Although its function is not fully understood, serum amyloid A1 appears to play a role in the immune system. Serum amyloid A1 may help repair damaged tissues, act as an antibacterial agent, and signal the migration of germ-fighting cells to sites of infection.

Levels of this protein increase in the blood and other tissues under conditions of inflammation. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight microbial invaders and facilitate tissue repair. When this has been accomplished, the body stops the inflammatory response to prevent damage to its own cells and tissues.

There are three versions of the serum amyloid A1 protein, known as alpha, beta, and gamma, which differ by one or two protein building blocks (amino acids). The frequency of these variants differs across populations. In white populations, the alpha version predominates and gamma is rare. In the Japanese population, however, the three versions appear almost equally.

Health Conditions Related to Genetic Changes

familial Mediterranean fever

Several studies of people with familial Mediterranean fever indicate that having the alpha version of the serum amyloid A1 protein increases the risk of a serious complication called amyloidosis. Amyloidosis involves the buildup of protein deposits that can lead to kidney failure if left untreated. Studies indicate that individuals with familial Mediterranean fever who also have the alpha version of the protein are two to seven times more likely to develop amyloidosis than are people with the beta or gamma version.

More serum amyloid A1 is produced in the body during episodes of inflammation such as those that occur in familial Mediterranean fever. This protein and related compounds may form abnormal clumps in the body's organs and tissues. It remains unclear, however, how the alpha version of serum amyloid A1 increases the susceptibility to amyloidosis (or alternatively, how the beta and gamma versions may protect against this complication) in people with this disorder.

other disorders

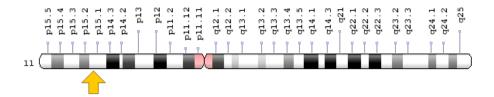
Among people with certain other inflammatory disorders, studies indicate that variants of the serum amyloid A1 protein also modify the risk of amyloidosis. For example, in the Japanese population, the gamma version of the protein appears to increase the risk of amyloidosis among adults with rheumatoid arthritis. Among white people with juvenile chronic arthritis, the alpha version indicates a high risk of developing amyloidosis.

More serum amyloid A1 is produced in the body during chronic inflammation such as occurs in these disorders. This protein and related compounds may form abnormal clumps in the body's organs and tissues. It remains unclear, however, how certain versions of serum amyloid A1 increase the susceptibility to amyloidosis.

Chromosomal Location

Cytogenetic Location: 11p15.1, which is the short (p) arm of chromosome 11 at position 15.1

Molecular Location: base pairs 18,266,225 to 18,269,977 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PIG4
- SAA
- SAA HUMAN
- TP53I4
- tumor protein p53 inducible protein 4

Additional Information & Resources

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SAA1%5BTIAB%5D%29+OR+%28serum+amyloid+A1%5BTIAB%5D%29%29+OR+%28PIG4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

 SERUM AMYLOID A1 http://omim.org/entry/104750

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC_SAA1.html
- HGNC Gene Family: Endogenous ligands http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc_data.php&hgnc_id=10513
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/6288
- UniProt http://www.uniprot.org/uniprot/P0DJI8

Sources for This Summary

- Bakkaloglu A, Duzova A, Ozen S, Balci B, Besbas N, Topaloglu R, Ozaltin F, Yilmaz E. Influence
 of Serum Amyloid A (SAA1) and SAA2 gene polymorphisms on renal amyloidosis, and on SAA/
 C-reactive protein values in patients with familial mediterranean fever in the Turkish population. J
 Rheumatol. 2004 Jun;31(6):1139-42.
 - Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15170927
- Ben-Chetrit E. Familial Mediterranean fever (FMF) and renal AA amyloidosis--phenotype-genotype correlation, treatment and prognosis. J Nephrol. 2003 May-Jun;16(3):431-4. Review.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12832747
- Booth DR, Booth SE, Gillmore JD, Hawkins PN, Pepys MB. SAA1 alleles as risk factors in reactive systemic AA amyloidosis. Amyloid. 1998 Dec;5(4):262-5.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10036584

- Cazeneuve C, Ajrapetyan H, Papin S, Roudot-Thoraval F, Geneviève D, Mndjoyan E, Papazian M, Sarkisian A, Babloyan A, Boissier B, Duquesnoy P, Kouyoumdjian JC, Girodon-Boulandet E, Grateau G, Sarkisian T, Amselem S. Identification of MEFV-independent modifying genetic factors for familial Mediterranean fever. Am J Hum Genet. 2000 Nov;67(5):1136-43. Epub 2000 Oct 3. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11017802
 Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1288556/
- Delibas A, Oner A, Balci B, Demircin G, Bulbul M, Bek K, Erdogan O, Baysun S, Yilmaz E. Genetic risk factors of amyloidogenesis in familial Mediterranean fever. Am J Nephrol. 2005 Sep-Oct;25(5): 434-40. Epub 2005 Aug 23.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16118480
- Gershoni-Baruch R, Brik R, Zacks N, Shinawi M, Lidar M, Livneh A. The contribution of genotypes at the MEFV and SAA1 loci to amyloidosis and disease severity in patients with familial Mediterranean fever. Arthritis Rheum. 2003 Apr;48(4):1149-55.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12687559
- Kelkitli E, Bilgici B, Tokgöz B, Dilek M, Bedir A, Akpolat I, Utas C, Akpolat T. SAA1 alpha/alpha alleles in amyloidosis. J Nephrol. 2006 Mar-Apr;19(2):189-91.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16736418
- Medlej-Hashim M, Delague V, Chouery E, Salem N, Rawashdeh M, Lefranc G, Loiselet J, Mégarbané A. Amyloidosis in familial Mediterranean fever patients: correlation with MEFV genotype and SAA1 and MICA polymorphisms effects. BMC Med Genet. 2004 Feb 10;5:4.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15018633
 Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC356915/
- Moriguchi M, Kaneko H, Terai C, Koseki Y, Kajiyama H, Inada S, Kitamura Y, Kamatani N. Relative transcriptional activities of SAA1 promoters polymorphic at position -13(T/C): potential association between increased transcription and amyloidosis. Amyloid. 2005 Mar;12(1):26-32.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16076608
- Moriguchi M, Terai C, Koseki Y, Uesato M, Nakajima A, Inada S, Nishinarita M, Uchida S, Nakajima A, Kim SY, Chen CL, Kamatani N. Influence of genotypes at SAA1 and SAA2 loci on the development and the length of latent period of secondary AA-amyloidosis in patients with rheumatoid arthritis. Hum Genet. 1999 Oct;105(4):360-6.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10543406
- Ray A, Shakya A, Kumar D, Benson MD, Ray BK. Inflammation-responsive transcription factor SAF-1 activity is linked to the development of amyloid A amyloidosis. J Immunol. 2006 Aug 15; 177(4):2601-9.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16888022
- OMIM: SERUM AMYLOID A1 http://omim.org/entry/104750
- Stevens FJ. Hypothetical structure of human serum amyloid A protein. Amyloid. 2004 Jun;11(2): 71-80.
 - Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15478462
- Thorn CF, Lu ZY, Whitehead AS. Tissue-specific regulation of the human acute-phase serum amyloid A genes, SAA1 and SAA2, by glucocorticoids in hepatic and epithelial cells. Eur J Immunol. 2003 Sep;33(9):2630-9.
 - Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12938239

- Utku U, Dilek M, Akpolat I, Bedir A, Akpolat T. SAA1 alpha/alpha alleles in Behçet's disease related amyloidosis. Clin Rheumatol. 2007 Jun;26(6):927-9. Epub 2006 Oct 13.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17039260
- Yamada T. Serum amyloid A (SAA): a concise review of biology, assay methods and clinical usefulness. Clin Chem Lab Med. 1999 Apr;37(4):381-8. Review.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10369107

Reprinted from Genetics Home Reference:

https://ghr.nlm.nih.gov/gene/SAA1

Reviewed: September 2008 Published: March 21, 2017

Lister Hill National Center for Biomedical Communications U.S. National Library of Medicine National Institutes of Health Department of Health & Human Services